Propionic Acidemia (PROP)

An organic acid disorder

What is it?

Propionic Acidemia (also known as PROP) is an inherited organic acid disorder. People with organic acid disorders, like PROP, cannot properly break down certain components of protein and fats. This is because the body is lacking a specific chemical called an enzyme. Since the body cannot properly break down the proteins and fats, certain organic acids build up in the blood and urine and cause problems when a person eats normal amounts of protein, or becomes sick.

What are the symptoms?

A person with PROP can appear normal at birth. Some people with PROP will have the following symptoms after a few days of life: poor feeding, lack of energy, vomiting, low muscle tone, and seizures. There is a high risk of infection, developmental delay, seizures, and neurological problems. Many symptoms of PROP can be prevented by immediate treatment and lifelong management. People with PROP typically receive follow-up care by a team of professionals that is experienced in treating people with metabolic disorders.

Inheritance and frequency

PROP is inherited in an autosomal recessive manner. This means that for a person to be affected with PROP, he or she must have inherited two non-working copies of the gene responsible for causing PROP. Usually, both parents of a person affected with an autosomal recessive disorder are unaffected because they are carriers. This means that they have one working copy of the gene, and one non-working copy of the gene. When both parents are carriers, there is a 1 in 4 (or 25%) chance that both parents will pass on the non-working copies of their gene, causing the baby to have PROP. Typically, there is no family history of PROP in an affected person. PROP is a rare fatty acid oxidation disorder; about 1 in 100,000 babies born have PROP.

How is it detected?

PROP may be detected through newborn screening. A recognizable pattern of elevated chemicals alerts the laboratory that a baby may be affected. Confirmation of newborn screening results is required to make a firm diagnosis. This is usually done by a physician that specializes in metabolic conditions, or a primary care physician.

How is it treated?

PROP is treated by eating a diet low in protein and drinking a special formula, and sometimes medication, as recommended by a genetic metabolic medical professional.

DISCLAIMER: This information is not intended to replace the advice of a genetic metabolic medical professional.

For more information:

Genetics Home Reference

Website: http://www.ghr.nlm.nih.gov

Save Babies Through Screening Foundation
4 Manor View Circle
Malvern, PA 19355-1622
Toll Free Phone: 1-888-454-3383
Fax: (610) 993-0545
Email: email@savebabies.org
Website: http://www.savebabies.org

Organic Acidemia Association
13210 - 35th Avenue North
Plymouth, MN 55441
Phone: 763-559-1797
Fax: 763-694-0017
Email: oaanews@aol.com
www.oaanews.org

American College of Medical Genetics
Newborn Screening ACT Sheets and Confirmatory Algorithms
http://www.acmg.net/resources/policies/ACT/condition-analyte-links.htm

Cardinal Glennon Children's Hospital
St. Louis, Missouri 314-577-5639
Website: http://pediatrics.slu.edu/index.phtml?page=geneticsdiv

Children's Hospital at University Hospital and Clinics
Columbia, Missouri 573-882-6991
Website: http://www.genetics.missouri.edu/

Children's Mercy Hospital
Kansas City, Missouri 816-234-3290
Website: http://www.childrens-mercy.org/content/view.aspx?id=155

St. Louis Children's Hospital
St. Louis, Missouri 314-454-6093
Website: http://www.peds.wustl.edu/genetics/